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Critical Analysis of Contemporary Public Health Genomics and Needs Assessment for Public
Health Genomics Course Curriculum

Kim London
Culminating Project Manuscript

Master in Public Health
Wright State University

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Table of Contents

Abstract	
Introduction.....	5
Statement of Purpose	7
Methods.....	8
Literature Review.....	9
General Current Status of Genomic Science	9
Personalized Medicine	12
Pharmacogenomics	16
Biobanks for Genetics.....	16
Environmental Genetics.....	18
Reproductive Genetic Testing.....	20
Assessment of Genomics in Public Health Education.....	22
Discussion	27
Discussion of Literature	27
Discussion of Genomics in Public Health Education	31
Recommendations and Conclusions	32
References.....	36
Appendix – List of Competencies Met in CE.....	43

Abstract

Objective: To analyze the need for increased content on genomics in Public Health (PH) academic curriculum.

Methods: A literature synthesis was performed of various genetic science studies. Results were assessed with regard to the number and type of genomic studies published, cost to perform genetic analyses, health care provider and the public's use and understanding of genetic testing capabilities. A search was conducted and assessment performed of public health genomics curriculum at accredited public health educational programs.

Results: The cost to sequence a single genome declined from \$10 million in 2007 to under \$10 thousand in 2012. This cost reduction correlates with an uptrend in published genome-wide association studies from about 50 published in 2007 to over 1300 published in 2012.

Approximately 2000 genetic tests are currently available with applications relevant to environmental genetics, personalized medicine and reproductive trait testing. A dearth of genetics-related material in academic PH programs is juxtaposed against current advancements in genetics.

Conclusions: Public health professionals should learn and develop genomic science materials for health promotion and education and policy addressing today's genetic determinants of health. A broad-based PH genomics course which introduces many issues versus deep details into a single genomics topic would address this need for the next generation of public health students and would be a valuable continuing education offering for current professionals. The discussion includes recommendations related to a full spectrum of PH programs, to include smaller programs that may not be in a position to add a new focus of study.

Keywords: genetics, DNA, personalized medicine, pharmacogenomics, academia

Critical Analysis of Contemporary Public Health Genomics and Needs Assessment for Public Health Genomics Course Curriculum

The National Human Genome Project and genetic science research is rapidly increasing a variety of uses for human genetic information in medical treatment, environmental genetics health risk assessments, and reproductive selection of viable embryos in fertility treatment. The Centers for Disease Control and Prevention has therefore dedicated efforts aimed at increasing activity and awareness in public health genomics (CDC, 2013). While some explorations in the human genetic sciences are in their infancy, this very fact brings uncertainties and anxieties that are already affecting human health in ways never before seen. For instance, a genetic test may indicate that an individual is susceptible to a particular disease, but a conundrum arises as to what to do with this knowledge without symptomatic condition or proven treatment options. Equal access to genetic tools is a concern that prevails in contemporary public health policy, parallel to the concern about equal access to other medical services. Without equal access to genetic services, social disparities in personal health, already a contemporary public health challenge, will be exacerbated.

Reproductive genetic technology also poses new public health policy concerns as rapidly advancing technology advances are used for reproductive gender selection and selection of non-diseased embryos for implantation. Broader application of these genetic technologies raise societal implications related to increasing gender choice and imbalances in health prosperity, which will compound existing public health challenges and could create new ones.

Environmental genomics is a related and compelling contemporary science that directly impacts public health policy. The aim of environmental genomics is to develop predictable correlation of response to environmental exposures by genetically susceptible persons. As this

science develops, it will allow for more precise disease/health risk determination and thus more targeted, less sweeping and less costly environmental health exposure regulations.

As genetic science capabilities continue to advance, guidelines for use of this information need to be reflected in public health practice and serve medical treatment modalities.

Accordingly, public health professionals should be on the front lines of elucidating and developing not only genomic science, but also the legal, ethical and social implications of genetic determinants of health.

Healthy People 2020 (United States Department of Health and Human Services [DHHS], 2013) contains an objective area for genomics, which was not in Healthy People 2010. Under the “Objectives” section of Healthy People 2020, several future opportunities of genomics in public health are recognized. These opportunities include specifically “[I]ncorporating health-related genomics education in primary, secondary, undergraduate, and graduate curricula” (DHHS, 2013, Emerging Issues in Genomics, line 7). This paper focuses on current graduate curricula and demonstrates a dearth of public health genomics materials in these programs. An introductory, broad topical public health genomics course (e.g., overview of many topics vs. deep detail into any one) would help fill this need initially for newly developing public health professionals and could be a valuable continuing education offering for established professionals.

Statement of Purpose

The purpose of this culminating experience manuscript is to determine whether public health academic programs include adequate curriculum related to current human genetics sciences. To this end, the manuscript will answer the following questions:

1. What is the state of genetic science at it relates to Public Health Genomics?

2. Does contemporary Public Health Genomics demonstrate a need to include more genomics-related material in standard Masters Public Health curricula?
3. What type of Public Health Genomics course would be useful to students' professional development?

Methods

A literature review and synthesis was performed of genetics-related peer-reviewed studies and other genetic science related professional source material. Studies and materials used were obtained using key word searches of “genetics,” “genomics,” and “public health”.

Quantifying analysis was used to illustrate increases in the number and type of genomic studies and decreases in cost to perform them. Prior survey research was assessed related to health care providers' use of genetics in clinical medicine and the public's use and understanding of genetic testing capabilities and results. An internet and literature search was conducted and critical content assessment performed of public health genomics resources and programs at academic public health programs in the United States. Findings were assessed with regard to applications of genetics and public health and corresponding existence of public health course curriculum.

Literature Review

General Current Status of Genomic Science

Healthy People 2020 is the first Healthy People program to include genomics in its targeted health outcomes. The goal is to “improve health and prevent harm through valid and useful genomic tools in clinical and public health practice” (DHHS, 2013, Goal, paragraph 1). Healthy People 2020 includes genomics because individuals' genetics play a role in nine out of ten leading causes of death, including heart disease, cancer, stroke, diabetes, and Alzheimer's disease (DHHS, 2013, Why Genomics is Important, paragraph 1). For example, studies show

that women who have inherited a disease-causing mutation in the BRCA1 gene have a 55 to 65 percent risk of developing breast cancer by age 70, and women who have inherited a disease-causing mutation in the BRCA2 gene have a 45 to 47 percent risk (Chen & Parmigiani, 2007; Antoniou, Pharoah, & Narod, 2003). For women whose genetic tests reveal the gene mutations for BRCA 1 and BRCA 2, breast surgery is an early intervention option that could reduce the risk of breast cancer by 95 percent (Hartman et al., 1999; Domchek et al., 2010; Rebbeck, Friebel, & Lynch, 2004; Meijers-Heijboer, Van Geel, & Van Putten, 2001).

The cost of genetic testing and the number of studies performed in recent years is relevant to the topic of the current state of genomic science. Figure 1 shows the cost of genetic sequencing since the completion of the Human Genome Project in 2003. The cost of genetic testing has been in a downward spiral since about 2008 (National Institutes of Health [NIH], 2013a). Figure 1 shows two metrics related to the cost of DNA sequencing. The "Cost per Megabase of DNA Sequence" is the cost of determining one megabase (Mb; a million bases) of DNA sequence. The "Cost per Genome" is the cost of sequencing a human-sized genome. The cost to sequence per human sized genome in 2007 was an estimated \$10 million; in 2012 the same test cost under \$10 thousand.

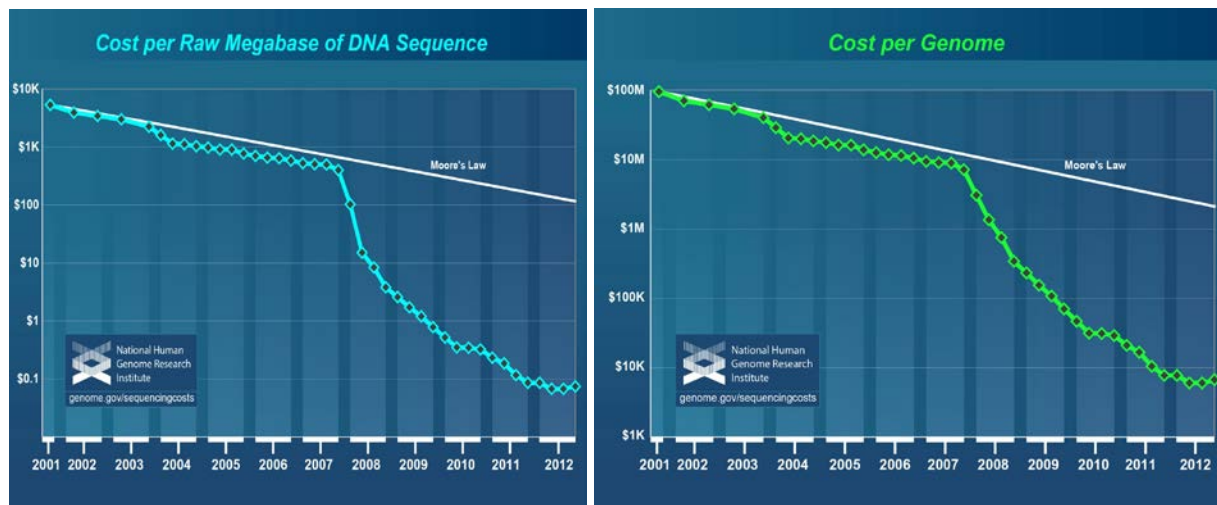


Figure 1. Cost of genetic sequencing trends.

Source: NIH, 2013b

The cost effectiveness of gene sequencing has been increasing over the last decade due to advance in technology such as automation. Prior to 2008, first generation sequencing platforms were used, and after 2008 new second generation sequencing platforms became available. These advancements have been driven by a competitive market to reduce costs to the point where genetic testing could be used in individual's routine medical care (Shendure, Mitra, Varma, & Church, 2004). Today, direct-to-consumer (DTC) genetic testing companies, such as *23andMe* (2013) provide personal genetic testing for \$99.00¹. The orders for these tests are placed entirely on-line at a computer with salivary DNA samples sent to the company via standard mail.

Figure 2 shows the total number of genome-wide research studies published from years 2004 to 2012. There is a notable upward trend beginning in 2008, the same year that the cost of genetic testing began to substantially decrease. Literature suggests there is an association between the decreased cost of genetic testing and the increase in genetics related studies. For example, Rowell (2013) conducted a study to determine trends in pathogen genetics studies and concluded that advances in technology have increased investigations into the role of human

¹ Further review of DTC genetic testing is addressed later.

genetic variation in the epidemiology of infectious diseases. Specifically, the study analyzed human genome epidemiology articles published from 2001 to 2010 (n=3,730), which included 23 genome-wide association studies (GWAS). The number of published articles each year increased from 148 in 2001 to 543 in 2010. Rowell (2013) concluded that as genomic research methods become more affordable, population-based research on infectious diseases will expand investigations into the role of variation in human genomes and bring new understanding of infectious disease susceptibility, severity, treatment, control, and prevention.

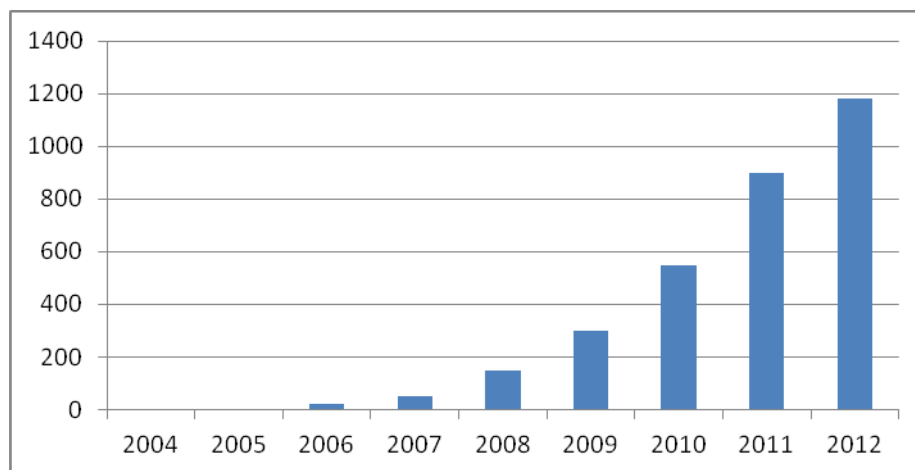


Figure 2. Genome-wide research studies published 2004-2012.

Source: NIH, 2013b

Personalized Medicine

Personalized medicine is the use of genetic testing to advance diagnosis of, or predictability for disease, and in turn provide earlier and individually tailored prevention and/or treatment intervention. Such tests are of interest to physicians, researchers, and members of the general public who are interested in better understanding their potential for developing specific diseases. It is therefore important for public health professionals to understand the changing landscape of tests available for this type of disease risk analysis.

There are about 2,000 genetic tests currently available for clinical use in personalized medicine efforts (CDC, 2013). However, Hamburg and Collins (2010) illustrates that there was no single public source of comprehensive information about these tests and whether they were cleared or approved by the United States Federal Drug Administration, which made it difficult for clinicians and consumers to make informed decisions about the testing to optimize individual health care. This problem was rectified on February 29, 2012, when the NIH started the Genetic Testing Registry (GTR), a centralized online resource for information about genetic tests (NIH, 2012). The intended audience for the GTR is health care providers and researchers.

Genetests is a medical genetic information resource owned by BioReference Laboratories created for physicians and geneticists that maintains a directory of laboratories offering genetic testing (Genetests.org, 2012). The organization reports exponential growth in the number of diseases tested for with molecular genetic testing. In 1993 there were approximately 100 clinics that tested for about 100 diseases. In 2012, just over 600 laboratories offered tests for over 2,900 diseases.

Three ways for an individual in the United States to obtain genetic testing include: as a patient from a clinical provider; as a research subject in a genetic research study; and, as a private consumer through commercial genetic testing companies. Each of these testing methods is reviewed below.

Clinical setting testing.

Despite the number of genetic tests increasingly available for clinical utility, studies indicate that health care providers may not be prepared to appropriately offering genetic tests or incorporating genetic testing in their clinical practice due to lack of familiarity or knowledge (Harvey et al., 2007; Cox et al., 2012). Harvey et al. (2007) reported that of 5,915 survey

respondents, 64% of patients with genetic conditions received no genetic information materials from their provider. Five years later, Cox et al. (2012) randomly surveyed 2,191 cancer providers in Oregon to determine genetic testing practices of these clinicians. Cox surveyed providers about their use of ten types of genetic cancer tests. The survey included a description of each genetic test and summary of the evidence-based recommendations published by the Evaluation of Genomic Applications in Practice and Prevention (EGAPP), the U.S. Preventive Services Task Force (USPSTF), and the National Comprehensive Cancer Network (NCCN). The results illustrated a lack of familiarity with the genetic test as the most common reason why clinicians did not order certain tests (Cox et al., 2012).

Genetic testing through research studies.

Various types of federally-funded genomic research studies are on-going in which thousands of individuals across the nation are recruited to be volunteer human research participants (National Human Genome Research Institute, 2013; Church, 2005; Coriell, 2011). Standards for genetic research are implemented by the NIH's Office of Human Research Protections (OHRP). Nonetheless, due to the breadth and pace of genetic science advanced, attendant future risks, clinical utility, and the full realm of ethical considerations are unknown (NHGRI, 2013; Church, 2005). At a minimum, today's genome testing is believed to carry the following possible risks to individuals: Infer paternity or other features of the participant's genealogy; possibility of statistical evidence that could affect employment or insurance or the ability to obtain financial services for the participant; reveal relatedness to criminals or incriminate relatives based on DNA samples used in forensic medicine; use of one's synthetic DNA at a crime scene; reveal propensity for a disease currently lacking effective treatment options (Caulfield, 2008; McGuire & Beskow, 2010; Church, 2005).

Direct-to-consumer testing.

Direct-to-consumer (DTC) genetic testing is commercial DNA testing that individuals may obtain without medical provider involvement and without medical indication. The tests are non-diagnostic and are marketed to provide insight to people about personal genetic traits and risk of disease and ancestry (CDC, 2013). These genetic tests are marketed on-line through various DTC company websites. The customer provides a salivary DNA sample to the company via regular mail. Examples of DTC companies are *Navigenics* (2013), *23andMe* (2013) and *deCODEme* (2013). Unlike DTC kits provided by the National Geographic Society (NGS) (2013) and Ancestry.com (2013) that are designed primarily to inform about probably ancestral groups, these companies offer genetic profile testing of hundreds of thousands of single nucleotide polymorphisms to provide consumers their personal risk of developing various disorders compared to the average population risk (Borry, 2010). Borry's history of the development of the DTC business sets forth that beginning in about 2007, advances in technology lowered the cost of testing and in turn proliferated the number of genome-wide association studies. However, there was little availability in the primary healthcare setting for testing so private companies began to offer this service to customers commercially. Borry (2010) reports that 2007 and 2008 saw a large number of DTC companies entered the market. The author reported finding 12 for-profit Internet companies offering limited susceptibility testing in 2003, and by 2009 there were over 30 companies offering some DTC services with a few offering whole genome sequencing (Borry, 2010). The DTC business brings concerns for potential consumers regarding credibility and comparability of tests, security of DNA use, privacy of genetic risk information, and lack of confidence in non face-to-face genetic counseling (Borry, 2010). Some DTC testing laboratories are voluntarily compliant with

Clinical Laboratory Improvement Amendments (CLIA) that establishes quality standards for laboratory testing (*23andMe*, 2013). However, there is no current Food and Drug Administration regulation that standardizes testing and test results analysis at DTC companies. Thus the value of the test results, in particular as they pertain to certain population sectors, remains uncertain (CDC, 2013; Aehnbauer, 2011; Borry, 2010).

Disparities in genetic testing.

The literature suggests concern for differential access to and utilization of genetic testing among racial and ethnic minorities compared to the majority white population, which may lead to compounded health disparities (Hall & Olopade, 2006; Pagan, 2009; Zehnbauer, 2011). Hall and Olopade (2006) report that genome testing research subjects come from a predominantly European descent; minority populations are therefore not well represented in genome reference bio-repositories that form the basis for diagnostic genetic testing.² Zehnbauer's (2011) report on DTC companies showed that DTC companies' interpretive associated risk reports are based upon published scientific research correlating particular genetic variants with a specific disease or condition; the majority of these peer-reviewed studies focused on Caucasian populations of European ancestry. Thus, relevance of the interpretation of the genetic tests is questionable for people of African, Asian, or Hispanic ancestry. Pagan (2009) analyzed a 2005 National Health Interview Survey (n=25,364), and showed a lack of awareness of genetic testing for cancer across racial and ethnic groups. Specifically, 48% of non-Hispanic whites reported that they heard about cancer genetic testing, while only 31% of blacks, 28% of Asians, and 19% of Hispanics were aware of genetic testing for cancer risk. Pagan concluded that culturally-tailored approaches are needed to improve awareness amongst minority groups to avoid further disparity

² Bio-repositories are reviewed on next page.

in cancer screening and outcomes. This type of health education disparity falls well in the domain of public health professionals.

Pharmacogenomics

Pharmacogenetics (PGx) is testing that analyzes a patient's genetic make-up to determine the best drug therapy for the particular patient based upon how they are expected to metabolize certain medications (NIHGR, 2013). The use of PGx is meant to improve the safety and effectiveness of drug therapies. Medco Health Solutions, Inc. and the American Medical Association (AMA) conducted a nation-wide survey of 10,000 physicians (Medco Health Solutions, Inc., 2009). Only 26% of the physicians reported any type of education in PGx and less than half of them (10%) believed they had the necessary information and training to put PGx testing to use in their clinical practice.

Bio-banks

In 2009 *TIME* magazine presented the "Top 10 Ideas Changing the World Right Now" (Park, 2009). Bio-banks was listed as number eight. Bio-banks (or repositories) are a collection of biospecimen samples often linked with individuals' demographic and/or health information to support a systematic approach to research. Biorepositories are not a new concept: skeletal collections and organ/tissue banks are more traditional examples of this practice. However, DNA bio-banks pose interesting research opportunities and ethical challenges as they are increasingly used for the developing field of genetic research.

There are numerous DNA bio-banks supporting genetic studies in the United States. For example, the Coriell Medical Institute Personalized Collaborative (CPMC) is a genetic study that includes DNA banking to advance personalized medicine and PGx (Coriell Institute for Medical Research, year). The CPMC study has 7,500 individual participants whose genetic data is linked

to their full medical and family history in the CPMC DNA databank. These thousands of DNA samples are banked for study purposes, and shared with other large DNA banks, such as NIH's Database of Genotypes and Phenotypes (dbGaP), a database at the National Center for Biotechnology Information designed to archive and distribute coded genotype, phenotype, exposure, and pedigree data from genome-wide association studies (DHHS, 2013). One of the largest bio-banks currently is from the Personal Genome Project (PGP) started at Harvard University (Church, 2005). This long-term project aims to sequence and publicize the complete genomes and (anonymized) medical records of 100,000 volunteers in order to help advance capabilities in personalized medicine.

Like other contemporary genetic banking studies, both CPMC and PGP are largely conducted via communications between volunteers and researchers using computer and the internet: this demonstrates how advances in technology drive larger databanks and make genetics studies easier for scientists to conduct. These same advantages bring challenging implications in data usage and access, data privacy and security and related implications. There remains no federal statutory guidelines on genetic bio-banks with the failure of the 2006 Genomics and Personalized Medicine Act (GovTrack, n.d.), which proposed to develop or expand population-based bio-banks to study genetic factors that influence drug efficacy and to develop usage guidelines for genetic bio-banks.

From the public health perspective, population-based bio-banking is a growing consideration. State health departments have recognized opportunities to use population-based bio-banks to be utilized to identify genes that contribute to human disease. For example, Connecticut's state health department's Virtual Office of Genomics stated that the its Departments of Public Health should be actively involved in public consultation and in

development of related legislation and infrastructure needed to support population-based bio-banks (Kelly, Stone, Manning, & Swede, 2007). Brand, Tobias, and Probst-Hensch (2012) discuss the use of genome-based bio-banking for public health research, surveillance systems, health policy development, individual health information management and effective health services. They report that public health's role with implications of research deriving from bio-banks should include establishing an epidemiological research agenda, balancing individual and social concerns, and promoting communication among genomics researchers, public health agencies, policymakers, and the public.

Environmental genetics.

The environmental genetics field of practice is concerned with the interaction between genes and the environment and the links to why some people get sick, while others do not (OPHG, 2013). Thus, environmental genetics is the link between the environment and human genes. Of the determinants of health, one's genetics is the least controllable component. However, there would be far reaching public health advantages to better predicting reactions to environmental exposures via genetic testing.

For example, building on an earlier study by Kalada (2006) and Ritz (2009) reports that pesticides plus certain human genetic types may increase risk of Parkinson's disease. This dopamine transporter genetic variant study found that people with a single susceptible allele who lived within 500 yards of fields where pesticides commonly used in agriculture³ were used had three times increased risk of developing Parkinson's disease. People who had two or more susceptible alleles had almost a 5-fold increase in risk. Ritz noted that people who were genetically susceptible but had no pesticide exposure showed no increased risk of Parkinson's. The study results suggested that individuals with a particular genetic make-up may be singularly

³The specific pesticides named in the study are maneb and paraquat.

sensitive to the neurodegenerative effects of certain pesticides; these findings could have implications for residents of agricultural communities as well as farm workers or industrial workers in plants that produce these pesticides.

Environmental health genetics emerged as a viable public health research area in the late 1990s. Kelada (2006) reported a list of seventeen proposed genetic effect modifiers of common exposures, which he concludes is suggestive of promising actionable findings with further research in these areas. Table 1 shows various studies that specifically assessed particular genotype interactions with pesticide exposures. All but one of these studies took place after the costs of genetic sequences began to decline in 2004, further evidence of the association between reduced cost of genetic sequence testing and increase in number of genetic studies. With the advances in genome-wide sequencing beginning in 2003, environmental health genetics has become more prominent in public health research. As the numbers and types of environmental health studies continue to grow, chronic disease may be better prevented in genetically high-risk populations.

Table 1.

Environmental Genetics Studies Pertaining to Pesticides and Genetic Type

Genotype Type	Exposure	Study
ABCB1	Organochlorine insecticides	Dutheil et al., 2010
MM PON1-55	Diazinon and Chlorpyrifos	Manthripragada et al., 2010
	Paraquat and Maneb	Ritz et al., 2009 confirming Kelada, 2006
P450 2D6	Pesticides	Elbaz et al., 2007
MnSOD	Pesticides	Fong et al., 2007
NQO1	Pesticides	Fong et al., 2007
Combined MnSOD/NQO1	Pesticides	Fong et al., 2007
	Paraquat and Maneb	Kelada et al., 2006
CYP 2D6 29B+ allele	Pesticides	Hubble, 1998

With the increasing development of environmental health genetics science, there are several related public health policy considerations. Resnik (2005) has identified several

developing policy areas related to public health genetics. They include: Should public health departments offer genetic tests? Should genetic tests only be offered to those in known environmentally risky areas? Should public health warn the public when predictive tests do evolve? Should product industry warn the public of possible genetic risks linked to their product? Should genetic tests be mandated for vulnerable groups, such as children? Should products be regulated differently regarding genetic links? Environmental genetics is a developing science with many major public health implications.

Reproductive genetic uses.

Preimplantation genetic diagnosis (PGD) is a means of detecting genetic disorders prior to an embryo being implanted (CDC, 2007). A single cell is biopsied from the embryos and tested before they are implanted. This allows for selection or de-selection of certain embryos prior to establishing pregnancy. PGD permits detection of genetic abnormalities and detection of certain early and late onset diseases. Thus, PGD helps reduce the number of newborns with or susceptible to specific genetic diseases, and avoids many issues related to considerations for termination of pregnancy in the later prenatal screening stages. However, PGD also allows for determination of other non-medical traits, such as gender, which introduces additional ethical considerations.

PGD is used by some to choose the sex of the embryo to be implanted as a means of family balancing. Generally, the use of PGD for testing of non-medical traits is controversial. Some countries that allow PGD also have legislature concerning PGD that prohibits sex selection (Viville & Pergament, 1998; Wells & Delhanty, 2001; Hudson, 2006; Baruch, 2008a). However, the United States permits non-medical PGD sex selection. There are no federal or state laws that directly regulate PGD for sex selection and professional guidelines, such as from the American

Society for Reproductive Medicine, are not binding (Deeney, 2013, p. 340). This leaves fertility clinics to devise their own policies on PGD. According to Hudson (2006) a 2004 survey study of almost five thousand Americans showed that nearly 50% of women and 35% of men approved of PGD testing for sex selection (p. 1642). Sidhu (2012) reported that \$18,000 was the average cost of a gender selection procedure at high-profile clinics with an estimated 4,000 to 6,000 procedures performed every year. “Fertility doctors foresee an explosion in sex-selection procedures on the horizon, as couples become accustomed to the idea that they can pay to beget children of the gender they prefer” (Sidhu, 2012, p.1).

A 2006 survey of 415 assisted reproductive technology (ART) clinics in the United States, showed that 93% of ART clinics that offered in-vitro fertilization (IVF clinics) provided PGD services to patients and 42% of these provided PGD for sex-selection in addition to health-related diagnosis (Baruch, 2008a). Baruch (2008a) concluded that PGD is widely provided by a large majority of United States IVF clinics, including PGD for sex selection.

Two other studies obtained direct data from IVF clinics pertaining to PGD outcomes for non-medical sex selection. Gleicherh and Barad (2007) obtained data from an IVF clinic in New York of 92 couples using PGD for family balancing between the years 2004-2006. A total of 56 male vs. 36 female embryos were selected for implantation. This study also included ethnicity and showed significantly higher male preference in Chinese, Arab/Muslim, and Asian-Indian, and for Western ethnicities a slight, but non-significant, preference for females. Finally, Colls (2009) obtained survey data from 246 clinics across the United States and found similar results with 127 male vs. 119 female embryos selected. Again, significant preferences for males were found in Chinese and Indian subjects, while a preference for females was found in Western subjects.

Related to reproduction and genetics, newborn genetic testing may become another area soon to be impacted by advancements in genetic science. The National Institutes of Health (NIH) recently published a news release stating that \$25 million dollars would be spent over the next five years in the Genomic Sequencing and Newborn Screening Disorders research program to explore possibilities of sequencing newborns' genomes. The goal is to determine whether useful medical information could be obtained by genetic testing beyond current newborn screening standards (National Institutes of Health [NIH], 2013c).

Assessment of Genomics in Public Health Education

Genomics in academia/public health education.

A Healthy People 2020 goal related to genomics recognizes several future opportunities of genomics in public health. Among these is: “[I]ncorporating health-related genomics education in primary, secondary, undergraduate, and graduate curricula” (DHHS, 2013, line 7).

In 1997, the CDC established the Office of Public Health Genomics (OPHG) that promotes the integration of genomics into public health research, policy, and practice (see www.cdc.gov/genomics). According to the OPHG, scientific developments over recent years have resulted in new potential for health impact. The 2011 OPHG Stakeholder Consultation Priorities Conference Report identifies advancing education in the public health profession regarding genomics as one of the 5 year priority items for 2012-2017 (Office of Public Health Genomics [OPHG], 2011, p. 17). The report found that there needs to be “a greater understanding by public health professionals of what genome-based knowledge can bring to public health practice” (p. 42). The report specifically recommends: “Incorporate genomics into the curricula of medical schools, nursing schools, and schools of public health” (p. 42). Thus, in 2012 OPHG developed five-year priorities for public health genomics as well as a specific action

plan for genomic implementation in public health practice (CDC, Public Health Genomics 2013 at a Glance, Section 1, n.d.).

In 2003 the Institute of Medicine (IOM) recommended that genetics be added as a new content area to be covered by every school of public health. This recommendation was made based upon IOM's assessment of the broader applications possible within public health of genomics versus previous genetic science aimed only at individuals. Recent personal correspondence with the three editors of the 2003 IOM report indicates the document has not been updated to determine whether and how the recommendations have been implemented (K. Gebbie, personal communication, 28 Oct. 2013; Rosenstock, 2013; L. Hernandez, personal communication, 28 Oct. 2013). Enough time has passed to make these assessments.

In 2007 the NIH's Secretary's Advisory Committee, Genetics, Health and Science (SACGHS) reported that there were 38 schools of public health in the United States that offered courses related to genetics or genomics. Of the 38 public schools, 11 had centers with concentrations in genetics or public health genomics and thus had several genetics and genomics related courses at these centers. The other 27 were general public health programs that offered one or two genetics related courses.

The literature review for this paper found no surveys or other data reporting the extent to which U.S. public health programs have incorporated genetics or genomics related courses since 2007. The Council on Linkages between Public Health Academia and Practice (housed as the Public Health Foundation) and the Association of Schools and Programs in Public Health (ASPPH) are two current organizations whose mission interests include public health core competencies and education of public health professionals. A search of these organizations' websites and published materials revealed no surveys of graduate public health education courses

related to genetics courses. In support of this paper, each of these organizations was asked to confirm whether they have conducted surveys of public health programs to obtain current data on how many programs offer genetics or genomics related courses and to what extent. The Council of Linkages did not respond. ASPHH responded that they have not and are not aware of any such data (Wiest, personal communication, 2013).

The Council on Education for Public Health (CEPH) is an independent agency recognized by the U.S. Department of Education to accredit schools of public health and public health programs outside schools of public health. On its website, CEPH (n.d.) lists the names of 100 of its certified public health programs. During this review, a random selection of twenty of the listed public health programs was analyzed for content regarding offering of genetics or genomics related courses. The twenty public health program 2013 curriculums were obtained from the respective programs' websites. Full course catalogs were not reviewed for alternate year courses were not accounted for in this review. A search of the 2013 course calendar was done for any course within the program that used the word "genetics" or "genomics" in the course title.

Table 2 shows the results of the program and curricula review. Of the twenty programs, none had a concentration in genetics or genomics and none had required courses in the area of genetics or genomics. Four of the ten offered one or two elective courses related to genetics or genomics: University of Maryland at Baltimore, University of Cincinnati, Oregon Health and Science University, and, New Mexico State University. While this assessment is not representative, it does provide some idea of the lack of genetic content in public health curricula.

Table 2***Twenty CEPH-accredited Public Health Programs and their Genetics or Genomics-related Courses***

Brown University Providence, RI	No mention of genetics or genomics in list of courses
East Carolina University Greenville, NC	No mention of genetics or genomics in list of courses
Eastern Kentucky University Richmond, KY	No mention of genetics or genomics in list of courses
Emory University Atlanta, GA	EH527 Biomarkers and Environmental Public Health
Jackson State University Jackson, MS	No mention of genetics or genomics in list of courses
Morgan State University Baltimore, MD	No mention of genetics or genomics in list of courses
New Mexico State University MPH Program in Community Health Education Las Cruces, NM	GERO 456. Biological Aspects of Aging. Aging, the developmental process of the body determined by cellular changes influenced by lifestyle, genetics, and environment. Investigates these changes, how health promotion influences them, and when they are considered a disease.
Northwestern Ohio Consortium for PH Toledo, OH	Genetic Epidemiology or Molecular Epidemiology
Oregon State University Portland, OR	PHPM 507 Genomics and Public Health: Current Issues and Future Trends in Healthcare and Policy
St. George's University Great River, NY	No mention of genetics or genomics in listed courses
San Diego State University San Diego, CA	No mention of genetics or genomics in listed courses
Temple University Philadelphia, PA	No mention of genetics or genomics in listed courses
University of Cincinnati Public Health Program Cincinnati, OH	PH 7064: Statistical Genetics (2013) PH 8056: Environmental Genetics and Molecular Toxicology
University of Florida Gainesville, FL	No mention of genetics or genomics in listed courses
University of Iowa Iowa City, IA	EPID5560 Introduction to Molecular Epidemiology EPID 5570 Genetics and Epidemiology
University of Kansas Kansas City, KS	No mention of genetics or genomics in list of courses
University of Maryland at Baltimore MPH Program Baltimore, MD	PREV 711, Genetic Epidemiology
University of West Florida Pensacola, FL	No mention of genetics or genomics in list of courses
University of Wisconsin-Madison Madison, WI	No mention of genetics or genomics in list of courses
Virginia Tech Blacksburg, VA	No mention of genetics or genomics in list of courses

Discussion

The completion of the National Human Genome Project, current advances in genetic testing technologies and the proliferation of scientific research are rapidly increasing the uses of human genetic information in medical treatment, reproductive applications, environmental genetics and genomics, as well as personal use of genetic testing through commercial enterprise. Because each of these areas has implications in public health, public health professionals should have working knowledge of this science and its implications. As indicated in the 2011 IOM recommendations, this knowledge should be made available in public health education programs.

Published genetic studies have exponentially increased since 2007 while the cost of genetic tests decreased. The association between the lowering cost of genetic testing and increase in published studies is reflected in the literature (Rowell, 2013; Borry, 2010; NIH, 2012). Consequently, genome-wide sequencing studies are being published and attendant data placed in evolving national genetic bio-repositories to more quickly standardize and advance genetic science. Resulting applications and opportunities are reflected in the literature. Examples are Rowell's (2013) report of expansions in infectious disease investigations, Borry's (2010) report of proliferation of consumer use of DTC testing, Ritz's (2009) report of environmental genomics applications, and documentation of increasing uses of genetic testing in reproduction (Hudson, 2006; Baruch, 2008b; Colls, 2009).

There is no indication that the trend of continued reduced cost, increases in research, and expanded use and commercializing of genetic testing is going to end. If this trend continues, genetics and genomics will change modern day medicine and broaden implications to public health.

With 2,000 (and counting) clinically relevant genetic tests available to clinicians, personalized medicine and pharmacogenomics have profound potential to prevent chronic disease, to save life, and to extend life expectancy—all of which are direct public health considerations (CDC, 2013). Early detection and intervention capabilities resulting from genetic tests could significantly improve success in preventive medicine, as was demonstrated in the BRCA 1 and BRCA 2 gene mutation, where multiple studies reported significant risk reduction for breast cancer (Hartman et al., 1999; Domchek et al., 2010; Rebbeck et al., 2004; Meijers-Heijboer et al., 2001).

Public health concerns related to personalized medicine include not only ensuring fair and affordable access to genetic testing, but also protection from possible harms of testing. The OPHG website offers public health community awareness initiatives that could help inform people about these risks, which may include: depression, anxiety, guilt, family tension, false sense of security, unclear results, costly testing and follow-up counseling (American Society of Clinical Oncology, 2012).

Cost of disease treatment after onset is a contemporary public health challenge that could be mitigated by genetics. It stands to reason the results of personalized medicine through genetics and pharmacogenomics would decrease cost of medical care by better targeting preventative treatments. Early detection and preventive treatment will result in fewer expressed diseases and disease sequelae. Tailored prescriptions can mean less trial and error with drugs, and thus less cost in pharmaceuticals.

The literature review in this paper supports Burke's six recommendations for the role of public health genomics in the "era of personalized medicine," for public health involvement in improving global human health through genomics:

- 1) Continue to integrate genomics into public health research and practice;
- 2) Establish and maintain appropriate research infrastructure for generating an evidence-base for genomic medicine;
- 3) Develop, implement, and evaluate model public health genomics programs and clinical services;
- 4) Promote international collaboration;
- 5) Foster appropriate genetic services and genome-based research;
- 6) Inform programs, research, and strategies in public health genomics by accepted ethical principles and practices (each cited verbatim from Burke et al., 2010, p. 789).

It is not possible for public health professionals to fulfill these roles if they are not properly educated in genomics and the public health applications.

Environmental genomics is a compelling contemporary science that directly impacts public health because its very aim is to develop predictable correlation of environmental exposures to genetically susceptible persons in order to allow for more precise risk determination and thus more precise (better targeted, less sweeping and less costly) environmental health exposure regulations. Rowell's 2013 study determined that trends in pathogen genetic studies revealing more knowledge about the role of human genetic variation in the epidemiology of infectious diseases. Kelada's (2006) and Ritz's (2009) findings related to exposures to pesticide linked to Parkinson's disease demonstrate the promise of future actionable preventative health potentials in environmental genomics applications. Kelada (2006) makes the case that more epidemiological studies are warranted to advance this science. If public health students are exposed early in their academic studies to this area of science, new epidemiologists may be more compelled to incorporate genetics into contemporary study designs. As genetic capabilities

continue to advance, it is imperative that guidelines for use of this information be reflected in public health practice. If science and related regulations are to advance, the public health work force must be educated in environmental genetics.

Reproductive genetic technology poses new significant public health policy concerns as technology is used for non-medical treatment purposes such as reproductive gender selection and selection of non-diseased embryos for implantation among those who can afford it. PGD for sex selection may be a valuable choice for family balancing, but is wrought with ethical implications yet to be resolved. Broader application of these genetic technologies may bring societal implications such as increasing gender choice and health imbalances, all of which will compound existing public health challenges and lead to new ones.

Contemporary genetic science brings public health issues related to equal access to genetic testing and genetic health awareness disparities. Without equal access to genetic services, social disparities in personal health, already a contemporary public health challenge will be exacerbated. The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) identified public genetics education as an important priority for public health professionals and recommends improvements in education related to genetics and genomics literacy (SACGHS, 2013). The literature review herein revealed areas where public health professionals should be concerned about growing disparities. Table 3 shows a compilation of areas where educated public health professionals could work to mitigate genetics and genomics related disparities. These areas include environmental and epidemiological research, public education on direct-to-consumer genetic services, facilitation of regulatory advancements on public accessibility to reproductive testing involving genetics, and developing and implementing culturally tailored awareness approaches related to genetics.

Table 3***Areas of Public Health Disparities Related to Genomics***

Research	Primarily uses white population as subjects. Results in lack of validity for clinical treatment, disease risk assessment, and environmental genomics applications across racial and ethnic groups. <u>Mitigation</u> : Educate epidemiologists and other genetic researchers to include subjects across racial and ethnic groups. Educate public health policy practitioners to implement and enforce research policy of fairness of representation in publically funded research.
Direct-to-Consumer Service	Socio-economic challenged may not afford the luxury of purchasing genetic tests commercially. <u>Mitigation</u> : Educate public health policy practitioners to implement fair public access to obtain genetic testing.
Reproductive Testing	Elective IVF services are expensive and not covered by medical insurance/welfare insurance. Socio-economic status will result in disparities of those obtaining reproductive testing for diseased embryos and/or trait selection. <u>Mitigation</u> : Educate public health policy practitioners to implement fair access regulations and usage of genetic reproductive technology.
Awareness	Nearly 50% of whites have heard of genetic testing, while 33% or lower of other racial and ethnic groups have not (Pegan, 2009). Lack of awareness among minority populations may lead to compounded health disparities. <u>Mitigation</u> : Educate public health community awareness practitioners about culturally tailored awareness approaches related to genetics.

Discussion of Genomics in Public Health Education

Professional organizations and national health agencies have repeatedly documented the needed to incorporate genetics and genomics in health education--not only medical and nursing education, but also public health academic education. The literature review provides ample documentation of leading national health organizations (NIH, IOM, CDC) that have determined it imperative for genetics to be included in public health academia. IOM's recommendation to include genomics in public health academia was published in 2003, yet it appears public health genetics related courses are still lacking from CEPH-accredited general public health graduate programs.

Studies support the need for increased genetics education amongst health professionals. Of 10,000 physicians surveyed nation-wide, only 26% of the physicians reported any type of education in pharmacogenetics and only 10% of physicians believed they had the necessary information and training to put pharmacogenetic testing to use in their clinical practice (Medco

Health Solutions, Inc., 2009). Over 60% of nearly 6,000 cancer patients surveyed were not provided genetic testing information from their providers (Harvey et al., 2007). As recently as 2012, 2,191 cancer doctors surveyed in Oregon did not perform validated/recommended cancer-related genetic tests for patients because they lacked familiarity with the tests themselves (Cox et al., 2012).

A barrier to including genetics in public health core curriculum may very well be crowded program curriculum in which genetics competes with other more obvious, well-known and popular public health concerns. Nonetheless, professional literature and studies repeatedly demonstrate the lack of genetics in public health education and recommend better inclusion of genetics in public health education to fight leading chronic diseases with genetic components and to prepare future public health professions for the advanced world of the genetic era.

Suggested Methods to Incorporate Genomics in Public Health Education

In order to assess the exact need today regarding gaps of genetics courses in public health programs, a comprehensive survey needs to be conducted to more precisely determine the extent to which today's public health students are exposed to genetics and genomics and related public health implications. From these data, public health must develop and incorporate additional genomics course material to fill the identified gaps in educational opportunities.

One method to influence inclusion of genetics and genomics material in public health academic programs is to clearly include genomics within core competencies for public health professionals. Academic programs are geared toward ensuring students are able to perform public health core competencies. If such core competencies include genomics, this in turn would motivate schools with public health programs to include more genomics materials in courses to better prepare students to fulfill the core competencies related to genomics.

A method to incorporate genetics and genomics material in the case of crowded public health curricula, would be to ensure relevant modules are included in existing core courses. The course title survey included in this article did not look at all material/modules within the courses offered, so some may already have inclusions related to genetics. A fuller survey of academic programs will solidify this. There are several possible ways to incorporate genetics in existing curricula because genetics and genomics relate to multiple aspects of traditional public health courses.

For instance, public health communication education and awareness courses could include methods of communicating awareness of complex and ever-evolving issues related to genetics. The health communication and awareness education should include aspects of disparities related to genetic research subjects and public genetic literacy and awareness of genetic testing across cultural lines. Public health professionals in the area of community education and awareness and global health are well positioned to advance awareness of genomics, decrease disparities, and increase appropriate genomic services that would save lives today (OPHG, 2013).

Another example is epidemiology courses could better serve future public health professionals if they included information about genome-wide association studies, the inclusion (or lack thereof) of minorities in such studies, and the Genetic Testing Registry (GTR) (the centralized online resource for information about genetic tests.) The intended audience for the GTR is health care providers and researchers (NIH, 2013d). Epidemiology courses are also a logical place to educate students about environmental genomics and inclusion of genetics in environmental health population studies. The new world of bio-repositories and population-based bio-banks is also ripe for inclusion in epidemiology education. Brand and colleagues

(2012) make a compelling case for use of genome-based biobanking for public health research and surveillance systems. Epidemiology students should learn that epidemiologists are in a good position to include genetics components in their research that can further advance the modern era of genomics for the betterment of public health.

Public health law, policy, management, and ethics courses could incorporate a plethora of genetic and genomics related materials to better educate students. Courses should educate students that public health professionals can and should be on the front lines of exploring and developing legal, ethical, and social implications of the genetic determinant of health and establishment of related policy. Policy education topics for genetics derived from the literature for this paper include: Fair access policy and services for genetic testing; environmental genomics regulation; ethical and legal standards for bio-repositories and use of genetic information; development of standards for genetic testing and analysis of test results, in particular from direct-to-consumer laboratories providing tests directly to the public without medical consultation; regulating use of pre-implantation genetic testing, in particular for late onset disease and trait testing.

Despite the value and need to include genetics in public health curriculum, the reality is that there are different types, sizes and levels of public health certificate and degree programs in the United States. Inclusion of additional material in the curriculum may be more challenging for smaller programs. Small public health education programs may not have the capacity to include more education material, let alone embark on courses on genomics or full concentrations on genomics. For these smaller programs, aspects of genomics could be incorporated into existing basic, required public health courses where appropriate. For instance, public health introduction courses could include a survey chapter on various aspects of public health

genomics. Environmental health courses could include more dedicated aspects of genomics and gene-environment interactions. Epidemiology courses and Global Health courses could include a lesson on genome-wide sequencing, the evolution of this science, and possible disparities of representative populations in epidemiological genetic studies. Health Communication and Awareness courses could include a chapter on the unique complexities of public health literacy regarding genetics and genomics, due partially to the quickly evolving/changing science. Public health law and policy basic courses could include a class on unique issues related to public health genomics evolving policy. Existing faculty need not have advanced knowledge in the area in order to ensure students are exposed to accurate public health genomics material. Instead, each of these classes could highlight the genomics related material by having an expert guest speaker in the genomics area discuss the topics in class.

Not all students need become public health genomics professionals, but all public health professionals and graduating students of public health programs should have a basic working understanding of the genetic sciences, today's applications, and future opportunities for genomics to improve public health. This basic professional competence could be obtained with some level of genomics material incorporated in all public health education programs.

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Appendix - List of Competencies Met in CE

Tier 1 Core Public Health Competencies

Domain #1: Analytic/Assessment
Describe the characteristics of a population-based health problem (e.g., equity, social determinants, environment)
Use methods and instruments for collecting valid and reliable quantitative and qualitative data
Identify sources of public health data and information
Identify gaps in data sources
Adhere to ethical principles in the collection, maintenance, use, and dissemination of data and information
Domain #2: Policy Development and Program Planning
Gather information relevant to specific public health policy issues
Describe how policy options can influence public health programs
Gather information that will inform policy decisions (e.g., health, fiscal, administrative, legal, ethical, social, political)
Domain #3: Communication
Identify the health literacy of populations served
Communicate in writing and orally, in person, and through electronic means, with linguistic and cultural proficiency
Participate in the development of demographic, statistical, programmatic and scientific presentations
Domain #4: Cultural Competency
Recognize the role of cultural, social, and behavioral factors in the accessibility, availability, acceptability and delivery of public health services
Domain #5: Community Dimensions of Practice
Identify stakeholders
Describe the role of governmental and non-governmental organizations in the delivery of community health services
Inform the public about policies, programs, and resources
Domain #6: Public Health Sciences
Identify the basic public health sciences (including, but not limited to biostatistics, epidemiology, environmental health sciences, health services administration, and social and behavioral health sciences)
Describe the scientific evidence related to a public health issue, concern, or, intervention
Retrieve scientific evidence from a variety of text and electronic sources
Discuss the limitations of research findings (e.g., limitations of data sources, importance of observations and interrelationships)
Partner with other public health professionals in building the scientific base of public health
Domain #7: Financial Planning and Management
N/A
Domain #8: Leadership and Systems Thinking
Describe how public health operates within a larger system
Identify internal and external problems that may affect the delivery of Essential Public Health Services
Participate in mentoring and peer review or coaching opportunities

Concentration Competencies

Health Promotion and Education:	
Area 1: Assess Needs, Assets and Capacity for Health Education	
1.2	Engage stakeholders to participate in the assessment process
Area 2: Plan Health Education Programs	
2.2	Select planning model(s) for health education
2.4	Formulate specific, measurable, attainable, realistic, and time-sensitive objectives
2.7	Organize health education into a logical sequence
Area 3: Implement Health Education	
3.1	Identify training needs
3.2	Develop training objectives
Area 4: Conduct Evaluation and Research Related to Health Education	
4.9	Disseminate research findings through professional conference presentations
Area 5: Manage Health Education Programs – N/A	
Area 6: Serve as a health education resource person	
6.6	Develop training plan
6.8	Use a variety of resources and strategies
6.10	Provide expert assistance
6.11	Evaluate the effectiveness of the expert assistance provided
Area 7: Communicate and advocate for health and health education	
7.1	Lead advocacy initiatives
7.4	Use evidence-based research to develop policies to promote health